AUTHOR INDEX TO VOLUME 22

Note: All notices for conferences, symposia, and meetings are under one heading: ANNOUNCEMENTS (Subject Index)

ABRISQUETA J A *see* RODRIGUEZ M T *et al*
ADŽIĆ S *see* MARKOVIĆ S *et al*
AFFARA N *see* CONNOR J M *et al*
WILCOX D E *et al*
AFFAZA N A *see* CONNOR J M *et al*
AKSOY M, KUTLAR A, KUTLAR F, DINÇOL G, ERDEM S, and BAŞTESBIHÇI S Survey on haemoglobin variants, β thalassaemia, glucose-6-phosphate dehydrogenase deficiency, and haptoglobin types in Turks from Western Thrace 288
AL-AWADI S A, TEEBI A S, FARAG T I, NAGUIB K M, and EL-KHALIFA M Y Profound limb deficiency, thoracic dystrophy, unusual facies, and normal intelligence: a new syndrome 36
ALLEN T D *see* HARRISON C J *et al*
ALLENSWORTH S *see* BERNAR J *et al*
ALTAAN M VAN *see* VAN ALTAAN M
ALTAY C and GURGEY A Clinical and haematological evaluation of β thalassaemia intermedia with increased Hb F and Hb A_2 in heterozygotes: β thalassaemia intermedia I 205
see also GURGEY A *et al*
ALVAREZ G *see* EVANS D A P *et al*
ANDERSON D C *see* GORDON M T *et al*
ANDERSON M A *see* HARPER P S *et al*
ARAD I *see* MENDELBERG A *et al*
ARIEL I *see* MENDELBERG A *et al*
AWADI S A AL- *see* AL-AWADI S A

BAKER S G, RABSON A, SHIRES R, JOFFE B I, and SEFTEL H C HLA antigens in South African Afrikaners with heterozygous familial hypercholesterolaemia: correspondence 320
BAKKER E *see* DEROM C *et al*
BALANZA E and TABOADA G The frequency of lactase phenotypes in Aymara children 128
BAMFORTH S *see* UPADHYAYA M *et al*
BANKIER A, DE CAMPO M, NEWELL R, ROGERS J G, and DANKS D M A pedigree study of perinatally lethal renal disease 104
BARAITSER M and PATTON M A Reduced penetrance in tuberous sclerosis 29
see also NORTH C *et al*
PATTON M A *et al*
SARGENT C *et al*
THOMPSON E *et al*
BARBIERI R *see* BERNARDI F *et al*
DEL SENNO L *et al*
BARNES I C S, KUMAR D, and BELL R J M A child with a recombinant of chromosome 8 inherited from her carrier mother: case report 67
BARREIRO C *see* SANCHEZ J M *et al*
BASS H N, SPARKES R S, LESSNER M M, FOX M, PHOENIX B, and BERNAR J A family with three independent autosomal translocations associated with 7q32→7qter syndrome 59
BAŞTESBIHÇI S *see* AKSOY M *et al*
BEITINS I Z *see* WILSON G N *et al*
BELL R J M *see* BARNES I C S *et al*
BERGHE H VAN DEN *see* VAN DEN BERGHE H
BERNAR J, SPARKES R S, and ALLENSWORTH S Interstitial deletion 2q24·3: case report with high resolution banding 226
see also BASS H N *et al*
BERNARDI F, DEL SENNO L, BARBIERI R, BUZZONI D, GAMBARI R, MARCHETTI G, CONCONI F, PANICUCCI F, POSITANO M, and PITRUZZELLO S Gene deletion in an Italian haemophilia B subject 305
see also DEL SENNO L *et al*
BERNSTEIN R, ISDALE J, PINTO M, DU TOIT ZAAIJMAN, and JENKINS T Short rib-polydactyly syndrome: a single or heterogeneous entity? A re-evaluation prompted by four new cases 46
BERRY A C *see* NEVILLE B G R *et al*
BERRY R J Centric fusions in Scottish mice 395 *abst*
BESTER A J *see* GROBLER-RABIE A F *et al*
BIANCO A *see* STABILE M *et al*
BIJLSMA J B *see* KLEP-DE PATER J M *et al*
BIKHAZI A B *see* BUHAZZA M A *et al*
BIRD T D *see* PAGON R A *et al*
BLANK C E *see* KUMAR D *et al*
BORGAONKAR D S Triple aneuploidy: correspondence 319
BORKOWSKA J *see* HAUSMANOWA-PETRUSEWICZ I *et al*
BOYD C D *see* GROBLER-RABIE A F *et al*
BOYD E *see* TOLMIE J L *et al*
BOYD Y, BUCKLE V J, and EDWARDS J H High resolution cytogenetic analysis of chromosomal translocations associated with Duchenne muscular dystrophy in females 139 *abst*
BRAMBATI B, SIMONI G, DANESINO C, OLDRINI A, FERRAZZI E, ROMITTI L, TERZOLI G, ROSSELLA F, FERRARI M, and FRACCARO M First trimester fetal diagnosis of genetic disorders: clinical evaluation of 250 cases 92
BREBNER D K *see* GROBLER-RABIE A F *et al*
BROCK D J H and VAN HEYNINGEN V Prenatal diagnosis of cystic fibrosis using monoclonal antibodies specific for the isoenzymes of alkaline phosphatase 139 *abst*

BROOK J D, SHAW D J, MEREDITH A L, HARLEY H G, SARFARAZI M, HUSON S M, and HARPER P S Localising the gene for myotonic dystrophy on chromosome 19 396 *abst*

see also HUSON S M *et al*

BROWN C *see* WILLIAMS H *et al*

BROWN C S, PEARSON P L, THOMAS N S T, SARFARAZI M, HARPER P S, and SHAW D J Linkage analysis of a DNA polymorphism proximal to the Duchenne and Becker muscular dystrophy loci on the short arm of the X chromosome 179

BUCKLE V J *see* BOYD Y *et al*

BUHAZZA M A, BIKHAZI A B, and KHOURI F P Evaluation of haematological findings in 50 Bahraini patients with sickle cell disease and in some of their parents 293

BUNDEY S, WEBB T P, THAKE A, and TODD J A community study of severe mental retardation in the West Midlands and the importance of the fragile X chromosome in its aetiology 258

BUNDEY S E *see* TUCKERMAN E *et al*

BUONOCORE M C *see* STABILE M *et al*

BURN J *see* SARGENT C *et al*

BUSH M *see* WILSON G N *et al*

BUZZONI D *see* BERNARDI F *et al*
DEL SENNO L *et al*

CADBURY R *see* THOMPSON E *et al*

CAINE A *see* TOLMIE J L *et al*

CAMPO M DE *see* DE CAMPO M

CANTÚ J M *see* RIVERA H *et al*

CAO A *see* MACCIONI L and CAO A

CHARLES N *see* DONNAI D *et al*

CHAY S O *see* TAY J S H *et al*

CHEAH K *see* SYKES B *et al*

CHITHAM R G *see* MARKS K *et al*

CIUFFO A A, CUNNINGHAM E, and TRAILL T A Familial pulmonary valve stenosis, atrial septal defect, and unique electrocardiogram abnormalities 311

CLARK B J *see* YATES J R W *et al*

CLARKE C A Lateral readings 10, 11, and 12: editorials, 1, 81, and 161

CLARKE M *see* YOUNG I D *et al*

CLEGG J B *see* WEATHERALL D J *et al*

CLINICAL GENETICS SOCIETY, meetings, November 1984, London, abstracts 137; April 1985, Birmingham, abstracts 393

COLE G *see* WILLIAMS H *et al*

COLOMBO B and FELICETTI L Admission of Hb S heterozygotes to a general hospital is relatively reduced in malarial areas 291

CONCONI F *see* BERNARDI F *et al*
DEL SENNO L *et al*

CONNEALLY P M *see* HARPER P S *et al*

CONNOR J M, PETTIGREW A F, HANN I M, FORBES C D, and AFFARA N Application of an intragenic genomic probe to genetic counselling for haemophilia B 138 *abst*

LOWE G D O, and AFFARA N A Application of an intragenic genomic probe to genetic counselling for haemophilia B in the west of Scotland 441

SHIACH C, HANN I M, and FORBES C D Clinical application of three intragenic DNA polymorphisms for carrier detection in haemophilia B 396 *abst*

CONWAY D I *see* GORDON M T *et al*

CORONA J SÁNCHEZ- *see* SÁNCHEZ-CORONA J

COSTER R VAN *see* VAN COSTER R

COUTURIER J *see* PLESSIS G *et al*

COUZIN D A *see* WATT J L *et al*

COX D W *see* HUGHES H E *et al*

CRAEN M *see* ORYE E *et al*

CUNNINGHAM E *see* CIUFFO A A *et al*

DALE R F The inheritance of primary lymphoedema 274

DALGLEISH R *see* GROBLER-RABIE A F *et al*

DALLAPICCOLA B, STOMEO C, FERRANTI G, DI LECCE A, and PURPURA M Discordant sex in one of three monozygotic triplets 6

DANESINO C *see* BRAMBATI B *et al*

DANKS D M *see* BANKIER A *et al*

DARLING S *see* GOODFELLOW P *et al*

DAVID T J and WINTER R M Familial absence of the pectoralis major, serratus anterior, and latissimus dorsi muscles 390

DAVIES K *see* UPADHYAYA M *et al*

DAVIES K E Molecular genetics of the human X chromosome: review article 243

see also PEMBREY M E *et al*
WINTER R M *et al*

DAVISON B C C *see* MUELLER R F *et al*

DE CAMPO M *see* BANKIER A *et al*

DE FRANCE H F *see* KLEP-DE PATER J M *et al*

DE LANGE G G *see* SANDERS P A *et al*

DE NEGROTTI T C *see* SANCHEZ J M *et al*

DE PATER J M KLEP- *see* KLEP-DE PATER J M

DEL SENNO L, PIRASTU M, BARBIERI R, BERNARDI F, BUZZONI D, MARCHETTI G, PERROTTA C, VULLO C, KAN Y W, and CONCONI F β^+-thalassaemia in the Po river delta region (northern Italy): genotype and β globin synthesis 54

see also BERNARDI F *et al*

DELAVENNE J *see* PLESSIS G *et al*

DEN BERGHE H VAN *see* VAN DEN BERGHE H

DEROM C, BAKKER E, VLIETINCK R, DEROM R, VAN DEN BERGHE H, THIERY M, and PEARSON P Zygosity determination in newborn twins using DNA variants 279

DEROM R *see* DEROM C *et al*

DESPOISSES S *see* PLESSIS G *et al*

DETTER J C *see* PAGON R A *et al*

DI LECCE A *see* DALLAPICCOLA B *et al*

DIEHL E J *see* TURKEL S B *et al*

DINÇOL G *see* AKSOY M *et al*

DOCHERTY Z and HULTÉN M A Extra euchromatic band in the qh region of chromosome 9: short report 156

DODGE A *see* ELLES R *et al*
DONNAI D, KILGORE C, KERZIN-STORRAR L, WATTERS E A, WEETMAN M, CHARLES N, and HARRIS R The emergency work of a genetic register 393 *abst*
DU TOIT ZAAIJMAN J *see* BERNSTEIN R *et al*
DUBOWITZ V *see* HILLIER J *et al*
DYER P A *see* SANDERS P A *et al*

EDWARDS J H Clinical application of linkage 139 *abst*
see also BOYD Y *et al*
EL-KHALIFA M Y *see* AL-AWADI S A *et al*
ELDER G *see* LAURENCE K M *et al*
ELLES R, KERZIN-STORRAR L, DODGE A, KILGORE C, READ A P, and HARRIS R Forty-four Duchenne muscular dystrophy families on a genetic register assessed with X chromosome probes 386 *abst*
EMERY A E H *see* YATES J R W and EMERY A E H
ERDEM S *see* AKSOY M *et al*
EVANS D A P, PATERSON S, FRANCISCO P, and ALVAREZ G The acetylator phenotypes of Saudi Arabian diabetics 479
see also SHAH R R *et al*
EVANS K T *see* LAURENCE K M *et al*

FARAG T I *see* AL-AWADI S A *et al*
FELICETTI L *see* COLOMBO B and FELICETTI L
FERGUSON-SMITH M A *see* WILCOX D E *et al*
YATES J R W *et al*
FERRANTI G *see* DALLAPICCOLA B *et al*
FERRARI M *see* BRAMBATI B *et al*
FERRAZZI E *see* BRAMBATI B *et al*
FITZSIMMONS J S Classroom teaching in genetics and birth defects: the Nottingham experience: annotation 345
and GUILBERT P R A family study of hidradenitis suppurativa 367
FLORES S A GONZÁLEZ- *see* GONZÁLEZ-FLORES S A
FORBES C D *see* CONNOR J M *et al*
FOX M *see* BASS H N *et al*
FRACCARO M *see* BRAMBATI B *et al*
FRANCE H F DE *see* DE FRANCE H F
FRANCISCO P *see* EVANS D A P *et al*
FREILIJ H *see* SANCHEZ J M *et al*

GAGE J P Clinical features of homozygous α2(I) collagen deficient osteogenesis imperfecta: correspondence 319
GAMBARI R *see* BERNARDI F *et al*
GARCIA M VARELLA- *see* VARELLA-GARCIA M
GARDINER A *see* OSCIER D G *et al*
GELLSTHORPE K *see* LAWRENCE J S *et al*
GIACOIA G P *see* SMITH V S and GIACOIA G P
GOODFELLOW P, DARLING S, and WOLFE J The human Y chromosome: review article 329
GONZÁLEZ-FLORES S A *see* RIVERA H *et al*
GORDON M T, CONWAY D I, ANDERSON D C, and HARRIS R Genetics and biochemical variability of variants of 21 hydroxylase deficiency 354
GRAHAM J M *see* THOMPSON E *et al*
GREGSON N M Stanley Walker, obituary 416
GRENNAN D M *see* SANDERS P A *et al*
GRIFFITHS B L *see* KUMAR D *et al*
GROBLER-RABIE A F, BREBNER D K, VANDENPLAS S, WALLIS G, DALGLEISH R, KAUFMAN R E, BESTER A J, MATHEW G G P, AND BOYD C D Polymorphism of DNA sequence in the human pro α2(1) collagen gene 182
GRUNEBAUM M *see* MERLOB P *et al*
GUILBERT P R *see* FITZSIMMONS J S and GUILBERT P R
GURGEY A, KAYIN S, KANSU E, and ALTAY C Clinical and haematological evaluation of β thalassaemia intermedia characterised by unusually low Hb F and increased Hb A_2: β thalassaemia intermedia II 213
see also ALTAY C and GURGEY A
GUSELLA J F *see* HARPER P S *et al*
GUSSON A C T *see* TAJARA E H *et al*
GUZMAN-RODRIGUEZ R *see* YATES J R W *et al*

HAMBLIN T J *see* OSCIER D G *et al*
HANCKE I and MILLER K Familial occurrence of a pseudodicentric chromosome 21: short report 155
HANN I M *see* CONNOR J M *et al*
HANSEN K L *see* YOUNG R S *et al*
HARDING A E, PETTY R K H, and MORGAN-HUGHES J A A mitochondrial myopathy: a genetic study 138 *abst*
HARLEY H G *see* BROOK J D *et al*
HARPER P S Gene mapping and medical genetics: editorial 241
Sir Cyril Clarke and Journal of Medical Genetics, an appreciation: editorial 417
YOUNGMAN, S, ANDERSON M A, SARFARAZI M, QUARRELL O, TANZI R, SHAW D, WALLACE P, CONNEALLY P M, and GUSELLA J F Genetic linkage between Huntington's disease and the DNA polymorphism G8 in South Wales families 447
see also BROOK J D *et al*
BROWN C S *et al*
HUSON S M *et al*
UPADHYAYA M *et al*
HARRIS R *see* DONNAI D *et al*
ELLES R *et al*
GORDON M T *et al*
HARRISON C J *et al*
SANDERS P A *et al*
HARRISON C J, JACK E M, ALLEN T D, and HARRIS R Investigation of human chromosome polymorphisms by scanning electron microscopy 16
HASEGAWA T *see* MOTEGI T *et al*
HAUSMANOWA-PETRUSEWICZ I, ZAREMBA J, and BORKOWSKA J Chronic proximal spinal muscular atrophy of childhood and adolescence: problems of classification and genetic counselling 350
HAYAKAWA K *see* OHDO S *et al*

HERR H M, HORTON S J, and SCOTT C I De novo paracentric inversion in an X chromosome: case report 140
HEYNINGEN V VAN *see* VAN HEYNINGEN V
HIBBARD B M *see* LAURENCE K M *et al*
HIDVÉGI J *see* SZABÓ M *et al*
HILL L *see* MARKS K *et al*
HILLIER J, JONES G E, STATHAM H E, WITKOWSKI J A, and DUBOWITZ V Cell surface abnormality in clones of skin fibroblasts from a carrier of Duchenne muscular dystrophy 100
HOOLE M *see* LAURENCE K M *et al*
HORTON S J *see* HERR H M *et al*
HOWARD P J *see* WALKER S *et al*
HUGHES H E, MCALPINE P J, COX D W, and PHILIPPS S An autosomal dominant syndrome with 'acromegaloid' features and thickened oral mucosa 119
HUGHES J A MORGAN- *see* MORGAN-HUGHES J A
HULTÉN M *see* LINDENBAUM R H *et al*
HULTÉN M A *see* DOCHERTY Z and HULTÉN M A
WALLACE B M N and HULTÉN M A
HUNTER D *see* WALKER S *et al*
HUSON S M, SHAW D J, MEREDITH A L, SARFARAZI M, BROOK J D, and HARPER P S Myotonic dystrophy, peripheral neurofibromatosis, and chromosome 19 markers 396 *abst*
see also BROOK J D *et al*

IANNUZZI S *see* STABILE M *et al*
IDLE J R *see* SHAH R R *et al*
ILCHYSYN A *see* WOODROW J C and ILCHYSYN A
ISDALE J *see* BERNSTEIN R *et al*

JACK E M *see* HARRISON C J *et al*
JAMES W H A possible mechanism underlying the sex selectivity of neural tube defect: correspondence 319
JENKINS T *see* BERNSTEIN R *et al*
JOFFE B I *see* BAKER S G *et al*
JOHNSTON A W *see* WATT J L *et al*
JONES G E *see* HILLIER J *et al*
JONGBLOET P H Periconceptional vitamin supplementation and the prevention of neural tube defects: correspondence 411
JUBERG R C, KNOPS J, and MOWREY P N Increased frequency of lymphocytic mitotic nondisjunction in recurrent spontaneous aborters 32

KAN Y W *see* DEL SENNO L *et al*
KANSU E *see* GURGEY A *et al*
KAUFMAN M H An hypothesis regarding the origin of aneuploidy in man: indirect evidence from an experimental model: hypothesis 171
KAUFMAN R E *see* GROBLER-RABIE A F *et al*
KAWAGUCHI K *see* OHDO S *et al*
KAYIN S *see* GURGEY A *et al*
KERZIN-STORRAR L *see* DONNAI D *et al*
ELLES R *et al*
KHALIFA M Y EL- *see* EL-KHALIFA M Y
KHOURI F P *see* BUHAZZA M A *et al*
KILGORE C *see* DONNAI D *et al*
ELLES R *et al*
KLEP-DE PATER J M, DE FRANCE H F, and BIJLSMA J B Interstitial deletion of the long arm of chromosome 11: case report 224
KNOPS J *see* JUBERG R C *et al*
KRSTIĆ M *see* MARKOVIĆ S *et al*
KRYWAWYCH S *see* PATTON M A *et al*
KUMAR D, BLANK C E, and GRIFFITHS B L Cornelia de Lange syndrome in several members of the same family 296
see also BARNES I C S *et al*
KUTLAR A *see* AKSOY M *et al*
KUTLAR F *see* AKSOY M *et al*

LABARTA J D *see* SANCHEZ J M *et al*
LANGE G G DE *see* DE LANGE G G
LAURENCE K M Four pregnancies with a male triploid fetus 137 *abst*
ELDER G, EVANS K T, HIBBARD B M, HOOLE M, and ROBERTS C J Should women at high risk of neural tube defect have an amniocentesis? 457
MORRIS J, and TEW B J The long term effects of a stillbirth with and without malformation on mothers and their families, 393 *abst*
see also PATTON M A and LAURENCE K M
LAUREYS G *see* ORYE E *et al*
LAWRENCE J S, GELLSTHORPE K, MORREL G, and MOLL J Genetic factors in ankylosing spondylitis 394 *abst*
LECCE A DI *see* DI LECCE A
LEE B W *see* TAY J S H *et al*
LEHMANN H *see* WORTHINGTON S and LEHMANN H
LEONARD J V *see* PEMBREY M E *et al*
LESSNER M M *see* BASS H N *et al*
LINDENBAUM R H, HULTÉN M, MCDERMOTT A, and SEABRIGHT M The prevalence of translocations in parents of children with regular trisomy 21: a possible interchromosomal effect? 24
LIPSON A H and WILLIAMS G Congenital diaphragmatic hernia in half sibs: case report 145
LOWE G D O *see* CONNOR J M *et al*

MCALPINE P J *see* HUGHES H E *et al*
MACCIONI L and CAO A Osmotic fragility test in heterozygotes for α and β thalassaemia 374
MCDERMOTT A *see* LINDENBAUM R H *et al*
MCEWAN H P *see* MORTIMER G *et al*
MCKUSICK V A Marcella O'Grady Boveri (1865–1950) and the chromosome theory of cancer 431
MCPHEAT J *see* POPE F M *et al*
MADOKORO H *see* OHDO S *et al*
MARCHETTI G *see* BERNARDI F *et al*
DEL SENNO L *et al*
MARKOVIĆ S, KRSTIĆ M, ŠULOVIĆ V, RADOJKOVIĆ Z and ADŽIĆ S Interstitial deletion of chromosome 2: short report 154

MARKS K, HILL L, CHITHAM R G, and WHITEHOUSE W L Interstitial deletion of chromosome 7p detected antenatally: case report 316
MARTIN M J *see* RODRIGUEZ M T *et al*
MATHEW C G P *see* GROBLER-RABIE A F *et al*
MEDRANO M A *see* YOUNG R S *et al*
MEHTA L and YOUNG I D Attitudes of Asian families to genetic counselling: correspondence 413
MELE B VAN *see* VAN MELE B
MENDELBERG A, ARIEL I, MOGLE P, and ARAD I Tracheo-oesophageal anomalies in the Goldenhar anomalad: case report 149
MEREDITH A L *see* BROOK J D *et al*
HUSON S M *et al*
MERLOB P, GRUNEBAUM M, and REISNER S H Familial opposable triphalangeal thumbs associated with duplication of the big toes: case report 78
see also SIVAN Y *et al*
MIGLIORINI A M *see* SANCHEZ J M *et al*
MILLER K *see* HANCKE I and MILLER K
MITTWOCH U Erroneous theories of sex determination: review article 164
MOGLE P *see* MENDELBERG A *et al*
MOLL J *see* LAWRENCE J S *et al*
MOLLER M *see* RIVERA H *et al*
MOORE J R *see* YOUNG I D and MOORE J R
MORGAN-HUGHES J A *see* HARDING A E *et al*
MORREL G *see* LAWRENCE J S *et al*
MORRIS J *see* LAURENCE K M *et al*
MORTIMER G, MCEWAN H P, and YATES J R W Fraser syndrome presenting as monozygotic twins with bilateral renal agenesis: case report 76
MOTEGI T, WATANABE K, NAKAMURA N, HASEGAWA T, and YANAGAWA Y De novo tandem duplication 9p (p12→p24) with normal GALT activity in red cells: case report 64
MOWREY P N *see* JUBERG R C *et al*
MUELLER R F, DAVISON B C C, and VAN ALTAAN M Practical considerations in the application of linked DNA polymorphisms in families at risk for Duchenne muscular dystrophy 137 *abst*
MUFTI G J *see* OSCIER D G *et al*

NAGUIB K M *see* AL-AWADI S A *et al*
NAKAMURA N *see* MOTEGI T *et al*
NEGROTTI T C DE *see* DE NEGROTTI T C
NEVILLE B G R, BERRY A C, and STODDART Y A case of malignant spinal cord ependymoma in association with a duplication of part of the long arm of chromosome 12: short report 154
NEVIN N C *see* SELLER M J and NEVIN N C
NEWELL R *see* BANKIER A *et al*
NICHOLLS A C *see* POPE F M and NICHOLLS A C
POPE F M *et al*
NISHIGUCHI T *see* OHDO S *et al*
NORTH C, PATTON M A, BARAITSER M, and WINTER R M The clinical features of the Cohen syndrome: further case reports 131

OATES N S *see* SHAH R R *et al*
OGILVIE D *see* SYKES B and OGILVIE D
OGILVIE D J *see* SYKES B C *et al*
OHDO S, MADOKORO H, SONODA T, NISHIGUCHI T, KAWAGUCHI K, and HAYAKAWA K Kabuki make-up syndrome (Niikawa-Kuroki syndrome) associated with congenital heart disease 126
OLD J M *see* PEMBREY M E *et al*
WEATHERALL D J *et al*
OLDRINI A *see* BRAMBATI B *et al*
OLSON I A *see* WATT J L *et al*
ORYE E, CRAEN M, LAUREYS G, VAN COSTER R, and VAN MELE B Extra Yq and partial monosomy 12p due to a Y;12 translocation in a boy with features of the 12p deletion syndrome: case report 222
OSCIER D G, MUFTI G J, GARDINER A, and HAMBLIN T J Reciprocal translocation between chromosomes 8 and 9 in atypical chronic myeloid leukaemia: case report 398
OWEN R *see* POPE F M *et al*

PAGON R A, BIRD T D, DETTER J C, and PIERCE I Hereditary sideroblastic anaemia and ataxia: an X linked recessive disorder 267
PANICUCCI F *see* BERNARDI F *et al*
PAPP Z *see* SZABÓ M *et al*
PATER J M KLEP-DE *see* KLEP-DE PATER J M
PATERSON C *see* SYKES B *et al*
PATERSON S *see* EVANS D A P *et al*
PATTON M A and LAURENCE K M Three new cases of oculodentodigital (ODD) syndrome: development of the facial phenotype 386
WINTER R M, KRYWAWYCH S, and BARAITSER M Raised 2 oxoglutarate in the DOOR syndrome 139 *abst*
see also BARAITSER M and PATTON M A
NORTH C *et al*
PEARSON P *see* DEROM C *et al*
PEARSON P L *see* BROWN C S *et al*
PEMBREY M *see* THOMPSON E and PEMBREY M
THOMPSON E *et al*
PEMBREY M E, OLD J M, LEONARD J V, RODECK C H, WARREN R, and DAVIES K E Prenatal diagnosis of ornithine carbamoyl transferase deficiency using a gene specific probe 462
see also SARGENT C *et al*
WINTER R M *et al*
PERROTTA C *see* DEL SENNO L *et al*
PETRUSEWICZ I HAUSMANOWA- *see* HAUSMANOWA-PETRUSEWICZ I
PETTIGREW A F *see* CONNOR J M *et al*
PETTY R K H *see* HARDING A E *et al*
THOMAS P K AND PETTY R K H
PHILIPPS S *see* HUGHES H E *et al*
PHOENIX B *see* BASS H N *et al*
PIERCE I *see* PAGON R A *et al*
PINTO M *see* BERNSTEIN R *et al*
PIRASTU M *see* DEL SENNO L *et al*
PITRUZZELLO S *see* BERNARDI F *et al*

PITT P and WILLIAMS I A new brachydactyly syndrome with similarities to Julia Bell types B and E 202
PLESSIS G, COUTURIER J, TURLEAU C, DESPOISSES S, and DELAVENNE J 'Pure' partial trisomy 2q in a male owing to malsegregation of a maternal translocation t(X;2)(p22·3;q32·1): case report 70
POPE F M and NICHOLLS A C Clinical features of homozygous α2(I) collagen deficient osteogenesis imperfecta: correspondence reply 319
McPHEAT J, and OWEN R Collagen genes and proteins in osteogenesis imperfecta 466
POSITANO M *see* BERNARDI F *et al*
PURPURA M *see* GALLAPICCOLA B *et al*

QUAIFE R *see* WILLIAMS T *et al*
QUARRELL O *see* HARPER P S *et al*

RABIE A F GROBLER- *see* GROBLER-RABIE A F
RABSON A *see* BAKER S G *et al*
RADOJKOVIĆ Z *see* MARKOVIĆ S *et al*
READ A P *see* ELLES R *et al*
REISNER S H *see* MERLOB P *et al*
SIVAN Y *et al*
RICHMOND J A *see* TURKEL S B *et al*
RICKETT A B *see* YOUNG I D *et al*
RIDDOCH D Adrenoleucodystrophy and adrenomyeloneuropathy 396 *abst*
RIVAS F *see* RIVERA H *et al*
RIVERA H, GONZÁLEZ-FLORES S A, RIVAS F, SÁNCHEZ-CORONA J, MOLLER M and CANTÚ J M Monosomy 13q32·3→qter: report of two cases: case report 142
ROBERTS C J *see* LAURENCE K M *et al*
RODECK C H *see* PEMBREY M E *et al*
RODRIGUEZ M T, MARTIN M J, and ABRISQUETA J A A complex balanced rearrangement involving four chromosomes in an azoospermic man: case report 66
RODRIGUEZ R GUZMAN- *see* GUZMAN-RODRIGUEZ R
ROGERS J G *see* BANKIER A *et al*
ROMITTI L *see* BRAMBATI B *et al*
ROSS H S *see* WATT J L *et al*
ROSSELLA F *see* BRAMBATI B *et al*

SANCHEZ J M, BARREIRO C, and FREILIJ H Two brothers with Martsolf's syndrome 308
LABARTA J D, DE NEGROTTI T C, and MIGLIORINI A M Complex translocation in a boy with trichorhinophalangeal syndrome: case report 314
SÁNCHEZ-CORONA J *see* RIVERA H *et al*
SANDERS P A, GRENNAN D M, DYER P A, DE LANGE G G, and HARRIS R Immunogenetic studies in families with rheumatoid arthritis and autoimmune thyroid disease 451
SARFARAZI M *see* BROOK J D *et al*
BROWN C S *et al*
HARPER P S *et al*
HUSON S M *et al*
UPADHYAYA M *et al*
WILLIAMS H *et al*
SARGENT C, BURN J, BARAITSER M, and PEMBREY M E Trigonocephaly and the Opitz C syndrome 39
SAUDER S E *see* WILSON G N *et al*
SCOTT C I *see* HERR H M *et al*
SEABRIGHT M *see* LINDENBAUM R H *et al*
SEFTEL H C *see* BAKER S G *et al*
SELLER M J Prenatal screening for Down syndrome: correspondence 321
and NEVIN N C Periconceptional vitamin supplementation and the prevention of neural tube defects: correspondence reply 411
SENNO L DEL *see* DEL SENNO L
SHAH R R, EVANS D A P, OATES N S, IDLE J R, and SMITH R L The genetic control of phenformin 4-hydroxylation 361
SHAW D *see* HARPER P S *et al*
SHAW D J *see* BROOK J D *et al*
BROWN C S *et al*
HUSON S M *et al*
SHENKIN A *see* YATES J R W *et al*
SHIACH C *see* CONNOR J M *et al*
SHIRES R *see* BAKER S G *et al*
SIMONI G *see* BRAMBATI B *et al*
SIVAN Y, MERLOB P, and REISNER S H Philtrum length, intercommissural distance. and ear measurements in newborn infants: correspondence 414
SMITH M A FERGUSON- *see* FERGUSON-SMITH M A
SMITH R *see* SYKES B *et al*
SMITH R L *see* SHAH R R *et al*
SMITH V S and GIACOIA G P Hyperinsulinaemic hypoglycaemia in an infant with mosaic trisomy 13: case report 228
SOLOMON E *see* SYKES B *et al*
SONODA T *see* OHDO S *et al*
SPARKES R S *see* BASS H N *et al*
BERNAR J *et al*
STABILE M, BIANCO A, IANNUZZI S, BUONOCORE M C, and VENTRUTO V A case of suspected teratogenic holoprosencephaly: case report 147
STATHAM H E *see* HILIER J *et al*
STEPHEN G S *see* WATT J L *et al*
STODDART Y *see* NEVILLE B G R *et al*
STOMEO C *see* DALLAPICCOLA B *et al*
STORRAR L KERZIN- *see* KERZIN-STORRAR L
ŠULOVIĆ V *see* MARKOVIĆ S *et al*
SUPER M, WATSON G H, and WRAITH E The velocardiofacial syndrome: fact or fantasy 393 *abst*
SWIFT P G F *see* YOUNG I D and SWIFT P G F
SYKES B and OGILVIE D Lethal osteogenesis imperfecta. Is the collagen gene deletion a normal variant rather than a causal mutation? 138 *abst*

SMITH R, VIPOND S, PATERSON C, CHEAH K, and SOLOMON E Exclusion of the α1(II) cartilage collagen gene as the mutant locus in type IA osteogenesis imperfecta 187
SYKES B C, OGILVIE D J, WORDSWORTH D P, and THOMPSON E Evidence against the structural gene encoding type II collagen (COL2A1) as the mutant locus in achondroplasia 394 *abst*
SZABÓ M, VARGA P, ZALATNAI A, HIDVÉGI J, TÓTH Z, and PAPP Z Sacrococcygeal teratoma and normal alphafetoprotein concentration in amniotic fluid: case report 405

TABOADA G *see* BALANZA E and TABOADA G
TAJARA E H, VARELLA-GARCIA M, and GUSSON A C T Partial trisomy 12q24·31→qter: case report 73
TANZI R *see* HARPER P S *et al*
TAY J S H, YIP W C L, YAP H K, LEE B W, WONG H B, and CHAY S O Dermatoglyphs in children with mitral valve prolapse 135
TAYLOR A M R Chromosomal studies as an aid in the diagnosis of ataxia telangiectasia 395 *abst*
TEEBI A S *see* AL-AWADI S A *et al*
TERZOLI G *see* BRAMBATI B *et al*
TEW B J *see* LAURENCE K M *et al*
THAKE A *see* BUNDEY S *et al*
THEIN S L *see* WEATHERALL D J *et al*
THIERY M *see* DEROM C *et al*
THOMAS N *see* UPADHYAYA M *et al*
WILLIAMS H *et al*
THOMAS N S T *see* BROWN C S *et al*
THOMAS P K and PETTY R K H Emery-Dreifuss muscular dystrophy 138 *abst*
THOMPSON E, CADBURY R, and BARAITSER M The Nager acrofacial dysostosis syndrome with the tetralogy of Fallot: case report 408
PEMBREY M Seckel syndrome: an overdiagnosed syndrome 192
Seckel syndrome and related disorders 139 *abst*
, and GRAHAM J M Phenotypic variation in LADD syndrome 382
see also SYKES B C *et al*
TODD J *see* BUNDEY S *et al*
TOLAROVA M Genetic investigation of cleft lip with or without cleft palate, a three generation family study of 578 pedigrees 137 *abst*
TOLMIE J L, CAINE A, and BOYD E The 11q− syndrome: six new cases 395 *abst*
TÓTH Z *see* SZABÓ M *et al*
TRAILL T A *see* CIUFFO A A *et al*
TUCKERMAN E, WEBB T, and BUNDEY S E Frequency and replication status of the fragile X, fra(X)(q27–28), in a pair of monozygotic twins of markedly differing intelligence 85
TURKEL S B, DIEHL E J, and RICHMOND J A Necropsy findings in neonatal asphyxiating thoracic dystrophy 112
TURLEAU C *see* PLESSIS G *et al*

UPADHYAYA M, BAMFORTH S, YOUNG I, THOMAS N, SARFARAZI M, DAVIES K, and HARPER P S Hunter's syndrome: evidence supporting a location on the distal part of the X chromosome long arm 394 *abst*
see also WILLIAMS H *et al*

VAN ALTAAN M *see* MUELLER R F *et al*
VAN COSTER R *see* ORYE E *et al*
VAN DEN BERGHE H *see* DEROM C *et al*
VAN HEYNINGEN V *see* BROCK D J H and VAN HEYNINGEN V
VAN MELE B *see* ORYE E *et al*
VANDENPLAS S *see* GROBLER-RABIE A F *et al*
VARELLA-GARCIA M *see* TAJARA E H *et al*
VARGA P *see* SZABÓ M *et al*
VENTRUTO V *see* STABILE M *et al*
VIPOND S *see* SYKES B *et al*
VLIETINCK R *see* DEROM C *et al*
VULLO C *see* DEL SENNO L *et al*

WAINSCOAT J S *see* WEATHERALL D J *et al*
WALKER S, HOWARD P J, and HUNTER D Familial complex autosomal translocations involving chromosomes 7, 8, and 9 exhibiting male and female transmission with segregation and recombination 484
WALLACE B M N and HULTÉN M A Meiotic chromosome pairing in the human female 395 *abst*
WALLACE P *see* HARPER P S *et al*
WALLIS G *see* GROBLER-RABIE A F *et al*
WARREN R *see* PEMBREY M E *et al*
WATANABE K *see* MOTEGI T *et al*
WATSON G H *see* SUPER M *et al*
WATT J L, OLSON I A, JOHNSTON A W, ROSS H S, COUZIN D A, and STEPHEN G S A familial pericentric inversion of chromosome 22 with a recombinant subject illustrating a 'pure' partial monosomy syndrome 283
WATTERS E A *see* DONNAI D *et al*
WEATHERALL D J, OLD J M, THEIN S L, WAINSCOAT J S, and CLEGG J B Prenatal diagnosis of the common haemoglobin disorders 422
WEBB T *see* TUCKERMAN E *et al*
WEBB T P *see* BUNDEY S *et al*
WEETMAN M *see* DONNAI D *et al*
WHITE M *see* YATES J R W *et al*
WHITEHOUSE W L *see* MARKS K *et al*
WILCOX D E, AFFARA N, YATES J R W, and FERGUSON-SMITH M A Linkage analysis of the X chromosome short arm leading to genetic counselling of the X linked muscular dystrophies using multiple flanking DNA polymorphisms 137 *abst*
WILLIAMS G *see* LIPSON A H and WILLIAMS G
WILLIAMS H, COLE G, THOMAS N, BROWN C, and SARFARAZI M Lethal X linked myotubular myopathy 138 *abst*

SARFARAZI M, UPADHYAYA M, BROWN C, and THOMAS N The use of 'flanking' markers to predict genotype in possible carriers and pregnancies at risk for X linked dystrophies 396 *abst*
WILLIAMS I *see* PITT P and WILLIAMS I
WILLIAMS T, ZARDAWI I, QUAIFE R, and YOUNG I D Complex cardiac malformation in a case of trisomy 9: case report 230
WILSON G N, SAUDER S E, BUSH M, and BEITINS I Z Phenotypic delineation of ring chromosome 15 and Russell-Silver syndromes: case report 233
WINTER R M The estimation of recurrent risks in monogenic disorders using flanking marker loci 12
PEMBREY M E, and DAVIES K E Analysis of linkage data with factor IX corroborates the hypothesis that a premutation, followed by a recombination event, generates the full mutation in fragile X linked mental retardation 397 *abst*
see also DAVID T J and WINTER R M
NORTH C *et al*
PATTON M A *et al*
WITKOWSKI J A *see* HILLIER J *et al*
WOLFE J *see* GOODFELLOW P *et al*
WONG H B *see* TAY J S H *et al*
WOODROW J C and ILCHYSYN A HLA antigens in psoriasis and psoriatic arthritis 492
WORDSWORTH D P *see* SYKES B C *et al*
WORTHINGTON S and LEHMANN H The first observation of Hb D Punjab β° thalassaemia in an English family with 22 cases of unsuspected β° thalassaemia minor among its members 377
WRAITH E *see* SUPER M *et al*
YANAGAWA Y *see* MOTEGI T *et al*
YAP H K *see* TAY J S H *et al*
YATES J R W and EMERY A E H A population study of adult onset limb-girdle muscular dystrophy 250
FERGUSON-SMITH M A, SHENKIN A, GUZMAN-RODRIGUEZ R, WHITE M, and CLARK B J Vitamin status and neural tube defects 394 *abst*
see also MORTIMER G *et al*
WILCOX D E *et al*
YIP W C L *see* TAY J S H *et al*
YOUNG I *see* UPADHYAYA M *et al*
YOUNG I D and MOORE J R Severe pseudoachondroplasia with parental consanguinity: case report 150
RICKETT A B, and CLARKE M High incidence of Meckel's syndrome in Gujarati Indians 301
and SWIFT P G F Parietal foramina in the Saethre-Chotzen syndrome: correspondence 413
see also MEHTA L and YOUNG I D
WILLIAMS T *et al*
YOUNG R S, MEDRANO M A, and HANSEN K L Partial 2p deletion in a girl with a complex chromosome rearrangement involving chromosomes 2, 6, 11, and 21: case report 401
YOUNGMAN S *see* HARPER P S *et al*
ZAAIJMAN J DU TOIT *see* DU TOIT ZAAIJMAN J
ZALATNAI A *see* SZABÓ M *et al*
ZARDAWI I *see* WILLIAMS T *et al*
ZAREMBA J *see* HAUSMANOWA-PETRUSEWICZ I *et al*

SUBJECT INDEX TO VOLUME 22

Note: All notices for conferences, symposia, and meetings are under one heading: ANNOUNCEMENTS

Abortion spontaneous, recurrent, increased frequency of lymphocytic mitotic non-disjunction 32
Abstracts, encouraging better standards 4 *abst*
Acetylator phenotypes, Saudi Arabian diabetics 479
Achondroplasia, structural gene encoding type II collagen (COL2A1) as mutant locus, evidence against 394 *abst*
'Acromegaloid' features and thickened oral mucosa, in autosomal dominant syndrome 119
Adrenoleucodystrophy and adrenomyeloneuropathy 396 *abst*
Adrenomyeloneuropathy and adrenoleucodystrophy 396 *abst*
Adult onset limb-girdle muscular dystrophy, population study 250
Alphafetoprotein concentration, normal, in anmniotic fluid, and sacrococcygeal teratoma: case report 405
Amniocentesis, for neural tube defect high risk 457
Amniotic fluid, normal alphafetoprotein concentration, and sacrococcygeal teratoma: case report 405
Aneuploidy
  origin in man, indirect evidence from experimental model: hypothesis 171
  triple: correspondence 319
Ankylosing spondylitis, genetic factors 394 *abst*
ANNOUNCEMENTS
  Clinical Genetics Society, meeting, Harrow, November 1985 328
  Prize 328
  Cystic Fibrosis Research Trust, Hon Medical Advisors' Conference, London, September 1985 160
  Genetics in Clinical Oncology, course 328
  International Symposium on Molecular Aspects of Neurobiology, Florence, June 1985 160
  Second International Symposium on the Fetus as a Patient: Diagnosis and Therapy, Jerusalem, May 1985 160
Asian families, attitudes to genetic counselling: correspondence 413
Asphyxiating thoracic dystrophy, neonatal, necropsy findings 112
Ataxia
  and hereditary sideroblastic anaemia, X linked recessive disorder 267
  telangiectasia, chromosomal studies as aid in diagnosis 395 *abst*
Atrial septal defect, familial pulmonary valve stenosis, and unique electrocardiogram abnormalities 311
Attitudes to genetic counselling, Asian families: correspondence 413
Autoimmune thyroid disease and rheumatoid arthritis, immunogenetic family studies 451
Autosomal dominant syndrome, with 'acromegaloid' features and thickened oral mucosa 119
Aymara children, frequency of lactase phenotypes 128

Becker muscular dystrophy
  chromosomal translocations, high resolution cytogenetic analysis 139 *abst*
  DNA polymorphisms 137 *abst*
  loci on chromosome X short arm, linkage analysis 179
Biographical details of contributors to special issue 419
Birth defects and genetics, classroom teaching, Nottingham experience: annotation 345
Blepharophimosis and female infertility: editorial 82
Bone marrow transplantation
  in cure of leukaemia 1 *abst*
  in mucopolysaccharidosis type VI syndrome: editorial 163
BOOK REVIEWS
  BENNETT P M and GRINSTED J *editors*: Plasmid technology. Methods in microbiology, volume 17, 1984 502
  BRACKEN M B *editor*: Perinatal epidemiology, 1984 500
  BUNDEY S Genetics and neurology, 1985 498
  COMMITTEE 5 REPORT. International Commission for Protection against Environmental Mutagens and Carcinogens. Perspectives in mutation epidemiology, 1984 158
  COX J G, COHEN B J, and LOEW F M *editors*: Laboratory animal medicine, 1984 501
  CRAWFORD M H *editor*: Black Caribs. A case in biocultural adaptation. Current developments in anthropological genetics, volume 3, 1984 499
  DE GROUCHY J and TURLEAU C Clinical atlas of human chromosomes, 2nd edition, 1984 238
  DEAN B J *editor*: Report of the UKEMS Sub-Committee on guidelines for mutagenicity testing, parts I and II, 1984 496
  EMERY A E H and RIMOIN D L *editors*: Principles and practice of medical genetics, volumes 1 and 2, 1983 238
  FINLEY S C, FINLEY W H, and FLOWERS C E *editors*: Birth defects: clinical and ethical considerations. Annual review of birth defects 1982, 1983 240

FREIRE-MAIA N and PINHEIRO M Ectodermal dysplasias: a clinical and genetic study, 1984 503
GOODFELLOW P *editor*: Genetic analysis of the cell surfaces 1984 324
GREEN M I and NISONOFF A *editors*: The biology of idiotypes, 1984 503
HAMERTON J L and FERGUSON-SMITH M A *editors*: Prenatal diagnosis. Special issue. Collaborative studies in prenatal diagnosis of chromosome aberrations, volume 4, 1984 237
HARDING A E The hereditary ataxias and related disorders, 1984 325
HARPER P S Practical genetic counselling 2nd ed. 1984 324
HARRIS H and HIRSCHHORN K *editors*: Advances in human genetics, volume 13, 1983 158
JONES D G Brave new people, 1984 502
KALTER H *editor*: Issues and reviews in teratology, volume 2, 1984 502
KENNETT R H, BECHTOL K B, and MCKEARN T J *editors*: Monoclonal antibodies and functional cell lines. Progress and applications, 1984 496
KINDT T J and CAPRA J D The antibody enigma, 1984 239
KIRSCH-VOLDERS M *editor*: Mutagenicity, carcinogenicity, and teratogenicity of industrial pollutants, 1984 326
LE GRYS V A The laboratory diagnosis of selected inborn errors of metabolism, 1984 497
LUDLOW C L and COOPER J A *editors*: Genetic aspects of speech and language disorders, 1983 325
MESSER A and PORTER I H *editors*: Recombinant DNA and medical genetics, 1983 158
ROMAN H L *editor*: Annual review of genetics, 1984 326
SASAZUKI T and TADA T *editors*: Immunogenetics. Its application to clinical medicine, 1984 497
SCHINZEL A Catalogue of unbalanced chromosome aberrations in man, 1984 237
SRŠEŇ S Alkaptonuria, 1984 159
The Harvey Lectures Series 78 (1982–1983), 1984 498
ZIMMERMAN B K Biofuture. Confronting the genetic era, 1984 499
Boveri, Marcella O'Grady (1865–1950), and the chromosome theory of cancer 431
Brachydactyly syndrome, new, similarities to Julia Bell types B and E 202

Cancer, chromosome theory, and Marcella O'Grady Boveri (1865–1950) 431
Carrier detection in haemophilia B, clinical application of three intragenic DNA polymorphisms 396 *abst*
Cell surface abnormality, in clones of skin fibroblasts from a Duchenne muscular dystrophy carrier 100
Centric fusions, in Scottish mice 395 *abst*
Cerebral haemorrhage, hereditary, γ trace alkaline microprotein: editorial 161
Cerebrotendinous xanthomatosis, long term treatment with chenodeoxycholic acid: editorial 162
Chenodeoxycholic acid, in long term treatment of cerebrotendinous xanthomatosis: editorial 162
Chromosome abnormalities, specific
2q, 'pure' partial trisomy, maternal translocation t(X;2)(p22·3;q32·1): case report 70
2, interstitial deletion: short report 154
2, 6, 11, and 21, and partial 2p deletion: case report 401
7, 8, and 9, familial complex autosomal translocations, involving, exhibiting male and female transmission with segregation and recombination 484
7p, interstitial deletion, detected antenatally: case report 316
7q32→7qter syndrome, with three independent autosomal translocations, in one family 59
8 and 9, reciprocal translocation, in atypical chronic myeloid leukaemia; case report 398
8, recombinant, inherited from carrier mother: case report 67
9, qh region, extra euchromatic band: short report 156
9p (p12→p24) tandem duplication, with normal GALT activity in red cells: case report 64
9 trisomy, with complex cardiac malformation: case report 230
11, long arm, interstitial deletion: case report 224
11q– syndrome, six new cases 395 *abst*
12, long arm, duplication and malignant spinal cord ependymoma: short report 154
12q24·31→qter, partial trisomy: case report 73
13, trisomy, in hyperinsulinaemic hypoglycaemia; case report 228
15 ring and Russell-Silver syndromes, phenotypic delineation: case report 233
19, myotonic dystrophy localisation 396 *abst*
19, myotonic dystrophy, and peripheral neurofibromatosis 396 *abst*
21, pseudodicentric, familial occurrence: short report 155
22, pericentric inversion, familial, recombinant subject, illustrating 'pure' partial monosomy syndrome 283
Chromosome abnormalities, general,
complex balanced rearrangement involving four chromosomes and azoospermia: case report 66
interstitial deletion 2q24·3: case report with high resolution banding 226
meiotic, pairing, human female 395 *abst*
monosomy 13q32·3→qter, report of two cases: case report 142
polymorphisms, human, investigation by scanning electron microscopy 16
theory of cancer, and Marcella O'Grady Boveri (1865–1950) 431
translocation in Duchenne and Becker muscular dystrophy, high resolution cytogenetic analysis 139 *abst*
Chromosome, X, de novo paracentric inversion: case report 140

X, fragile fra(X)(q27–28) frequency and replication status, twins 85
linkage data with factor IX 397 *abst*
, and severe mental retardation, West Midlands, community study 258
X, long arm, location of Hunter's syndrome 394 *abst*
, transposition to Y short arm during human evolution: editorial 81
X, molecular genetics: review article 243
X probes and Duchenne muscular dystrophy 396 *abst*
X short arm, Duchenne or Becker muscular dystrophy, DNA polymorphisms 137 *abst*
Duchenne and Becker muscular dystrophy loci, linkage analysis of DNA polymorphism proximal 179
Y, human: review article 329
Y;12 translocation, resulting in extra Yq and partial monosomy, 12p deletion syndrome: case report 222
Chronic proximal spinal muscular atrophy, of childhood and adolescence, classification and genetic counselling 350
Clarke, Sir Cyril, and Journal of Medical Genetics: an appreciation: editorial 417
Cleft lip, with or without cleft palate, genetic investigation, three generation family study of 578 pedigrees 137 *abst*
Clinical Genetics Society meetings, Birmingham, April 1985, abstracts 393, London, November 1984, abstracts 137
Cohen syndrome, clinical features, further case reports 131
Collagen genes
deletion, lethal osteogenesis imperfecta, normal variant or causal mutation 138 *abst*
and proteins, osteogenesis imperfecta 466
Collagen type II (COL2A1), as mutant locus in achondroplasia, evidence against 394 *abst*
Congenital heart disease, and Kabuki make-up syndrome (Niikawa–Kuroki syndrome) 126
Cornelia de Lange syndrome, in several members of the same family 296
CORRESPONDENCE
A possible mechanism underlying the sex selectivity of neural tube defect 319
Attitudes of Asian families to genetic counselling 413
Clinical features of homozygous α2(I) collagen deficient osteogenesis imperfecta 319
HLA antigens in South African Afrikaners with heterozygous familial hypercholesterolaemia 320
Parietal foramina in the Saethre-Chotzen syndrome 413
Periconceptional vitamin supplementation and the prevention of neural tube defects 411
Philtrum length, intercommissural distance, and ear measurements in newborn infants 414
Prenatal screening for Down syndrome 321
Triple aneuploidy 319
Cystic fibrosis
gene, searching for 2 *abst*
improved sweat test method for diagnosis: editorial 83
prenatal diagnosis using monoclonal antibodies specific for isoenzymes of alkaline phosphatase 139 *abst*

Deletion, 2p, partial, with complex chromosome rearrangement involving chromosomes 2, 6, 11, and 21: case report 401
Dermatoglyphs, in children with mitral valve prolapse 135
Diabetics, acetylator phenotypes, Saudi Arabia 479
Diaphragmatic hernia, congenital, in half sibs: case report 145
DNA polymorphisms
Duchenne or Becker muscular dystrophy 137 *abst*
G8 and Huntington's disease, genetic linkage between, South Wales families 447
proximal to Duchenne and Becker muscular dystrophy loci on chromosome X short arm, linkage analysis 179
three intragenic, clinical application for carrier detection in haemophilia B 396 *abst*
DNA sequence, in human pro α2(I) collagen gene, polymorphism 182
DNA variants, zygosity determination, newborn twins 279
DOOR syndrome, and raised 2 oxoglutarate 139 *abst*
Down syndrome, prenatal screening: correspondence 321
Duchenne muscular dystrophy
cell surface abnormality in clones of skin fibroblasts from a carrier 100
chromosomal translocations, high resolution cytogenetic analysis 139 *abst*
DNA polymorphisms 137 *abst*
forty-four families on genetic register assessed with X chromosome probes 396 *abst*
loci on chromosome X short arm, linkage analysis of DNA polymorphism proximal 179
use of 'flanking' markers to predict genotype in possible carriers and pregnancies at risk 396 *abst*

Ear measurements, newborn infants: correspondence 414
Embryos, developing, patterns 1 *abst*
Emery-Dreifuss muscular dystrophy 138 *abst*
Ependymoma, spinal cord, malignant, and chromosome 12 long arm duplication: short report 154

Factor VIII gene, characterisation: editorial 81
Factor IX and fragile X, linkage data 397 *abst*
Familial hypercholesterolaemia,
see Hypercholesterolaemia
Fetal diagnosis, first trimester, genetic disorders, clinical evaluation of 250 cases 92
Fraser syndrome, presenting as monozygotic twins with bilateral renal agenesis: case report 76

GALT normal activity in red cells, with tandem duplication 9p (p12→p24): case report 64

Gamma trace alkaline microprotein and hereditary cerebral haemorrhage: editorial 161
Gene
deletion, in Italian haemophilia B subject 305
mapping and medical genetics: editorial 241
therapy, a step nearer 3 *abst*
Genetics
and birth defects, classroom teaching, Nottingham experience: annotation 345
counselling, attitudes of Asian families: correspondence 413
for haemophilia B, application of intragenic genomic probe, West Scotland 441
register, emergency work 393 *abst*
Glucose-6-phosphate dehydrogenase deficiency, survey, Turks from Western Thrace 288
Goldenhar anomalad, tracheo-oesophageal anomalies: case report 149
Gujarati Indians, high incidence of Meckel's syndrome 301

H-Y antigen, lack, male sexual differentiation, mice: editorial 161
Haemoglobin
disorders, common, prenatal diagnosis 422
S heterozygotes, admission to general hospital, reduced in malarial areas 291
variants, survey, Turks from Western Thrace 288
Haemophilia B
genetic counselling for, application of intragenic genomic probe, West Scotland 441: 138 *abst*
clinical application of three intragenic DNA polymorphisms for carrier detection 396 *abst*
Italian subject, gene deletion 305
Haptoglobin types, survey, Turks from Western Thrace 288
Hidradenitis suppurativa, family study 367
HLA antigens
in psoriasis and psoriatic arthritis 492
in South African Afrikaners with heterozygous familial hypercholesterolaemia: correspondence 320
Holoprosencephaly, teratogenic, suspected: case report 147
Homoeosis, sequences of genes and mutant occurrences 1 *abst*
Hunter's syndrome, location on distal part of X chromosome long arm, evidence supporting 394 *abst*
Huntington's disease and DNA polymorphism G8, genetic linkage between, South Wales families 447
21 hydroxylase deficiency, genetics and biochemical variability of variants 354
Hypercholesterolaemia
familial 1 *abst*
heterozygous South African Afrikaners, HLA antigens: correspondence 320
severe, liver transplantation to provide low density lipoprotein receptors: editorial 163
Hyperinsulinaemic hypoglycaemia, and mosaic trisomy 13: case report 228

Immunogenetic studies, in families with rheumatoid arthritis and autoimmune thyroid disease 451
Infectious disease, changing pattern: editorial 83
Infertility
female, and blepharophimosis: editorial 82
and sperm defects: editorial 82
Intercommissural distance, newborn infants: correspondence 414
Interstitial deletion, chromosome 7p, detected antenatally: case report 316
Intragenic genomic probe, application to genetic counselling for haemophilia B 138 *abst*

Kabuki make-up syndrome (Niikawa-Kuroki syndrome), and congenital heart disease 126

Lacrimo-auriculo-dento-digital syndrome, phenotypic variation 382
Lactase phenotypes, frequency in Aymara children 128
LADD syndrome *see* Lacrimo-auriculo-dento-digital syndrome
Lateral readings 10, 11, and 12: editorials 1, 81, 161
Latissimus dorsi, pectoralis major, and serratus anterior muscles, familial absence 390
Lesch-Nyhan syndrome, first trimester diagnosis: editorial 84
Leukaemia
adult acute non-lymphocytic, high resolution chromosomes as independent prognostic indicator: editorial 82
bone marrow transplantation in cure of 1 *abst*
chronic myeloid, atypical, reciprocal translocation of chromosomes 8 and 9: case report 398
Limb deficiency, profound, thoracic dystrophy, unusual facies, normal intelligence, a new syndrome 36
Linkage
analysis of DNA polymorphism proximal to Duchenne and Becker muscular dystrophy loci on chromosome X short arm 179
clinical application 139 *abst*
Low density lipoprotein receptors, liver transplantation to provide, in severe hypercholesterolaemia: editorial 163
Lung cancer, metabolic oxidation phenotypes as markers for susceptibility: editorial 162
Lymphocytic mitotic non-disjunction, increased frequency in recurrent spontaneous abortion 32
Lymphoedema, primary, inheritance 274

Malarial areas, admission of Hb S heterozygotes to general hospital reduced 291
Marker loci, flanking, use to estimate recurrence risks in monogenic disorders 12
Martsolf's syndrome, in two brothers 308
Meckel's syndrome, high incidence in Gujarati Indians 301
Meiotic chromosome pairing, human female 395 *abst*
Melanoma, prevention by recognition of its precursors: editorial 161

Mental retardation, severe, and fragile X chromosome, West Midlands, community study 258
Metabolic oxidation phenotypes, as markers for lung cancer susceptibility: editorial 162
Mitochondrial myopathy, genetic study 138 *abst*
Mitral valve prolapse, children, dermatoglyphs 135
Molecular genetics, X chromosome: review article 243
Monoclonal antibodies for isoenzymes of alkaline phosphatase, in prenatal diagnosis of cystic fibrosis 139 *abst*
Monogenic disorders, estimation of recurrence risks using flanking marker loci 12
Monosomy
  13q32·3→qter, report of two cases: case report 142
  syndrome, 'pure' partial, familial pericentric inversion of chromosome 22 with recombinant subject illustrating 283
Mucopolysaccharidosis type VI syndrome, bone marrow transplantation: editorial 163
Muscular
  atrophy, spinal, chronic proximal form, of childhood and adolescence, classification and genetic counselling 350
  dystrophy, adult onset limb-girdle, population study 250
  see also Becker: Duchenne
Myopathy, mitochondrial, genetic study 138 *abst*
Myotonic dystrophy
  localisation, chromosome 19 396 *abst*
  peripheral neurofibromatosis, and chromosome 19 markers 396 *abst*
Myotubular myopathy, X linked, lethal 138 *abst*

Nager acrofacial dysostosis syndrome, with tetralogy of Fallot: case report 408
Narcolepsy, genetic markers: editorial 84
Neural tube defect
  amniocentesis for women at risk 457
  possible mechanism underlying sex selectivity: correspondence 319
  prevention with periconceptional vitamin supplementation: correspondence 411
  vitamin status 394 *abst*
Niikawa-Kuroki syndrome, and congenital heart disease 126
Nottingham, classrooom teaching in genetics and birth defects: annotation 345
Nuclear transplantation, mammals, methods and success 1 *abst*

Obituary Stanley Walker, 416
Oculodentodigital (ODD) syndrome, three new cases, development of facial phenotype 386
Opitz C syndrome, and trigonocephaly 39
Oral mucosa, thickened, and 'acromegaloid' features, in autosomal dominant syndrome 119
Ornithine carbamoyl transferase deficiency, prenatal diagnosis using gene specific probe 462
Osmotic fragility test, in heterozygotes, for α and β thalassaemia 374
Osteogenesis imperfecta 1 *abst*
  collagen genes and proteins 466
  homozygous α2(I) collagen deficient, clinical features: correspondence 319
  lethal, collagen gene deletion, normal variant or causal mutation 138 *abst*
  type IA, exclusion of α 1(II) cartilage collagen gene as mutant locus 187
Oxoglutarate, 2, raised, in DOOR syndrome 139 *abst*

Parietal foramina, in Saethre-Chotzen syndrome: correspondence 413
Pectoralis major, serratus anterior, and latissimus dorsi muscles, familial absence 390
Periconceptional vitamin supplementation, and prevention of neural tube defects: correspondence 411
Peripheral neurofibromatosis, myotonic dystrophy, and chromosome 19 markers 396 *abst*
Phenformin 4-hydroxylation, genetic control 361
Philtrum length, newborn infants: correspondence 414
Pluripotent haematopoietic stem cells, introduction of new genetic material, mouse 3 *abst*
Po river delta region (northern Italy), β^+- thalassaemia, genotype and β globin synthesis 54
Polymorphisms, chromosome, human, investigation by scanning electron microscopy 16
Pregnancy, trophoblast cells in peripheral blood: editorial 81
Prenatal diagnosis
  common haemoglobin disorders 422
  cystic fibrosis, using monoclonal antibodies specific for isoenzymes of alkaline phosphatase 139 *abst*
  ornithine carbamoyl transferase deficiency, using gene specific probe 462
  see also Down syndrome
Pro α1(II) cartilage collagen gene, exclusion as mutant locus in type IA osteogenesis imperfecta 187
Pro α2(I) collagen gene, polymorphism of DNA sequence 182
Probe
  gene specific, for prenatal diagnosis of ornithine carbamoyl transferase deficiency 462
  intragenic genomic, application to genetic counselling for haemophilia B in West Scotland 441
Pseudoachondroplasia, severe, with parental consanguinity: case report 150
Psoriasis and psoriatic arthritis, HLA antigens 492
Pulmonary valve stenosis, familial, atrial septal defect, and unique electrocardiogram abnormalities 311

Renal
  agenesis, bilateral, monozygotic twins, and Fraser syndrome: case report 76
  disease, perinatally lethal, pedigree study 104
  restriction fragment length polymorphism (RFLP) and Duchenne muscular dystrophy carriers 137 *abst*
  in Duchenne and Becker muscular dystrophy 179
  pro α2(I) collagen gene 182
Rheumatoid arthritis and autoimmune thyroid disease, immunogenetic studies in families with 451

Ring chromosome 15 and Russell-Silver syndromes, phenotypic delineation: case report 233
Risks, recurrence, in monogenic disorders estimation using flanking marker loci 12
Russell-Silver and ring chromosome 15 syndromes, phenotypic delineation: case report 233

Sacrococcygeal teratoma, and normal alphafetoprotein concentration, in amniotic fluid: case report 405
Saethre-Chotzen syndrome, and parietal foramina: correspondence 413
Saudi Arabia, diabetics, acetylator phenotypes 479
Seckel syndrome
overdiagnosed 192
and related disorders 139 *abst*
Serratus anterior, pectoralis major, and latissimus dorsi muscles, familial absence 390
Sex determination, erroneous theories: review article 164
Short rib-polydactyly syndrome, single or heterogeneous entity, re-evaluation 46
Shoulder girdle muscles, familial absence 390
Sickle cell disease, evaluation of haematological findings in 50 Bahraini patients and some parents 293
Sideroblastic anaemia, hereditary, and ataxia, X linked recessive disorder 267
South Wales families, genetic linkage between Huntington's disease and DNA polymorphism G8 447
Sperm defects and infertility: editorial 82
Spinal
cord ependymoma, malignant, and chromosome 12 long arm duplication: short report 154
muscular atrophy, chronic proximal form, of childhood and adolescence, classification and genetic counselling 350
Stillbirth, with and without malformation, long term effects on mothers and their families 393 *abst*
Sweat test, improved method for diagnosis of cystic fibrosis: editorial 83

Tetralogy of Fallot, and Nager acrofacial dysostosis syndrome: case report 408
Thalassaemia
α and β, osmotic fragility test in heterozygotes 374
β, survey, Turks from Western Thrace 288
β^+, Po river delta region (northern Italy), genotype and β globin synthesis 54
Hb D Punjab β^o, first observation in an English family with 22 cases of unsuspected β^o thalassaemia minor among its members 377
intermedia β, with increased Hb F and Hb A_2 in heterozygotes, clinical and haematological evaluation: part I β thalassaemia intermedia I 205: part II β thalassaemia intermedia II 213
Thumbs, triphalangeal, familial opposable, and duplication of big toes: case report 78
Toes, big, duplication, and familial opposable triphalangeal thumbs: case report 78
Tracheo-oesophageal anomalies, Goldenhar anomalad: case report 149
Translocation
autosomal, familial complex, involving chromosomes 7, 8, and 9, exhibiting male and female transmission with segregation and recombination 484
independent three, in one family, associated with 7q32→7qter syndrome 59
chromosomal, in Duchenne and Becker muscular dystrophy, high resolution cytogenetic analysis 139 *abst*
chromosomes 8 and 9, in atypical chronic myeloid leukaemia: case report 398
complex, in trichorhinophalangeal syndrome: case report 314
see also Chromosomes: Trisomy
Transplantation, nuclear, mammals, methods and success 1 *abst*
Trichorhinophalangeal syndrome, and complex translocation: case report 314
Trigonocephaly, and Opitz C syndrome 39
Triple aneuploidy: correspondence 319
Triplets, monozygotic, one with discordant sex 6
Triploid fetus, male, in four pregnancies 137 *abst*
Trisomy
2q, partial, 'pure', owing to maternal translocation t(X;2)(p22·3;q32·1):case report 70
12q24·31→qter: case report 73
9, with complex cardiac malformation: case report 230
13, mosaic, and hyperinsulinaemic hypoglycaemia: case report 228
21, translocations, prevalence in parents of children with 24
Trophoblast cells, in peripheral blood, pregnancy 81
Tuberous sclerosis, reduced penetrance 29
Turner's syndrome in one of three triplets 6
Twins
chromosome fragile X, fra(X)(q27–28) frequency and replication status 85
monozygotic, with bilateral renal agenesis, presenting with Fraser syndrome: case report 76
newborn, zygosity determination using DNA variants 279

Velocardiofacial syndrome, fact or fantasy 393 *abst*
Vitamin
status, neural tube defects 394 *abst*
supplementation, periconceptional, and prevention of neural tube defects: correspondence 411

Wilson's disease, effective treatment with oral zinc sulphate 4 *abst*

X linked
myotubular myopathy, lethal 138 *abst*
recessive disorder hereditary sideroblastic anaemia and ataxia 267

Zinc sulphate, oral, Wilson's disease treatment 4 *abst*
Zygosity determination, newborn twins, using DNA variants 279